

Case Report

Penile hypoplasia associated with anorectal malformation: What should we call this condition?

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ABSTRACT

Background: Urogenital abnormalities are common in patients with anorectal malformations (ARM). The wide spectrum of urogenital abnormalities and ARM sometimes make it difficult to name the conditions simply.

Case Presentation: We experienced a case of penile hypoplasia associated with the urethrorectal communication.

Conclusion: From the viewpoint of urogenital anomalies, we might call this condition “aphallia variant” or “absent penis spectrum”. On the other hand, from the viewpoint of ARM, we might call this condition “male posterior cloaca” or “partial urorectal septum malformation sequence”. More important than naming them is understanding the clinical conditions precisely.

Keywords: Anorectal malformations, Aphallia, Cloaca.

INTRODUCTION

Urogenital abnormalities are often associated with anorectal malformations (ARM) with a reported incidence of 30% to 50%.[1] The wide spectrum of urogenital abnormalities and ARM sometimes makes it difficult to name the conditions simply. We report a case of penile hypoplasia associated with ARM that we puzzled over naming.

CASE REPORT

A baby boy was delivered at 34 weeks gestation by Cesarean section due to worsening oligohydramnios. His birth weight was 2364g. Antenatal ultrasonography showed bowel dilatation at 20 weeks gestation. He had respiratory distress at birth, and he needed to be on a ventilator for a short period. The abdomen was remarkably distended. The anus was located in the normal position. The abdominal distention improved after catheterization through the anus. Ultrasonography showed the distended bladder with bilateral hydroureteronephrosis. The urethral meatus could not be found. On day 1, a suprapubic tube was placed in the bladder, and bilateral hydroureteronephrosis was

resolved. On day 6, cystography via the suprapubic tube suggested a rectovesical fistula without detecting the anterior urethra. Because catheterization through the anus was absolutely required for relieving abdominal distention, a colostomy was created on day 15. At 7 months of age, he was referred to our center for further management.

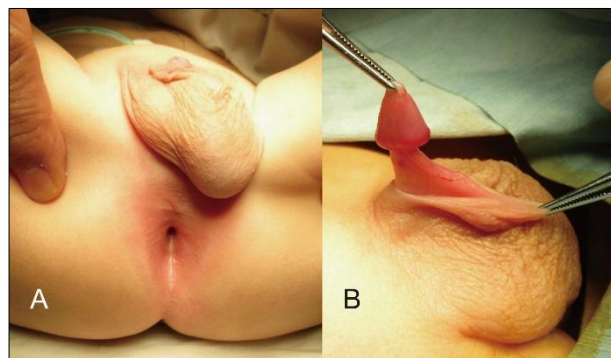


Figure 1: A) A protrusion of the genital skin like a phallus was noted. The anus was located in the normal position. B) Genital examination under general anesthesia showed only the glans without the corporeal bodies or urethral meatus

A protuberance of the genital skin like a phallus with a small glans was found, but the corporeal bodies were not palpated (Fig. 1A). MRI showed only the crus without normal corporeal bodies (Fig. 2). It also showed no spinal abnormalities. Distal colonography revealed a distended rectum. The left with compensatory hypertrophy was positioned the scrotum, but the right testis was not palpated.



Figure 2: A) MRI showed only the crus without normal corporeal bodies. B) A catheter was placed in the bladder through the urethrorectal communication.

At 9 months of age, genital examination under general anesthesia revealed only the glans without the corporeal bodies or urethral meatus (Fig. 1B). Additionally, the anal canal was so narrow that the little finger could not be inserted into it. Endoscopy through the anus showed the urethral opening was located at the anterior wall of the rectum, 2 cm from the anal verge, and the normal caliber urethra was 5 cm in length. The prostatic utricle, which was 1 cm in depth, was found in the posterior urethra. Subsequently, a vesicostomy was created, and the right testis was resected because inguinal exploration confirmed it was the nubbin. The histopathological study showed the resected nubbin contained no viable testicular tissue.

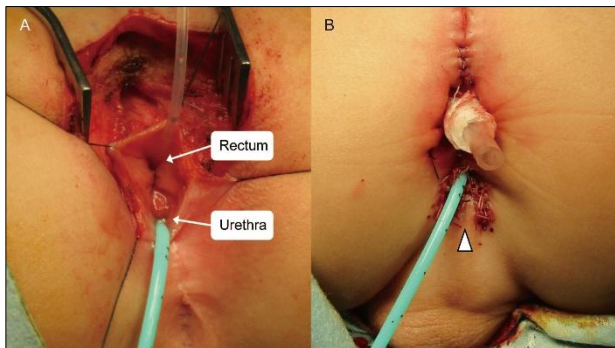


Figure 3: (A) The rectum and urethra were mobilized via a posterior sagittal approach. (B) The urethra was separated from the rectum and mobilized anteriorly. Perineal urethrotomy was created using the skin flap.

Based on the above findings, it was concluded that he had penile hypoplasia with anorectal malformation. At 19 months of age, posterior sagittal anorectoplasty was performed. At the same time, the urethra was anteriorly mobilized, and perineal urethrotomy was created with the skin flap (Fig. 3). Postoperative cystography via

vesicostomy confirmed he was able to void well without residual urine through the perineal meatus. At 2 years of age, cystoscopy and urethral calibration performed in conjunction with closure of the colostomy confirmed the intact meatus and urethra. We are planning to close the vesicostomy at around 3 years of age. We are also taking into consideration urethral reconstruction with preputial skin and buccal mucosal graft in the future.

DISCUSSION

Urogenital anomalies are common in patients with ARM. Understanding urogenital anomalies is very important due to their impact on the overall morbidity and mortality of these patients.[1] In the upper urinary tract, the most common abnormalities are renal dysplasia /agenesis, vesicoureteral reflux, hydronephrosis, renal duplications, and renal ectopia. Associated abnormalities of the lower urinary and genital tract include hypospadias, epispadias/exstrophy, urethral strictures, or urethral duplications.[2] Both urogenital and anorectal anomalies display a wide spectrum in these patients. Therefore, we sometimes puzzle over what we should call these complicated conditions. We experienced a case of penile hypoplasia associated with a urethrorectal communication.

From the viewpoint of urogenital anomalies, we might be able to call this condition “aphallia variant” or “absent penis spectrum”.[3] Aphallia is a rare abnormality with an estimated incidence of 1 in 10 million to 30 million births resulting in complete or partial development failure of the genital tubercle. Aphallia is characterized by absence of the corpora cavernosa and corpus spongiosum.[4] Rudimentary erectile tissue is occasionally present. In the case series of Hendren, one patient had a rudimentary flaccid structure without the corpora, a small glans with a tiny opening at the tip, and a thread-like urethra that ended blindly. That patient also had an enlarged prostatic utricle like our case.[5] Similarly, Coquet-Reinier et al. reported an atypical case of aphallia with a rudimentary genital bud and atrophic corpora cavernosa.[6]

From the viewpoint of anorectal malformations, we might be able to call this condition “male posterior cloaca”[3] or “partial urorectal septum malformation sequence (partial URSMS)”.[7,8] A cloaca is a malformation that makes an impact on the rectum and urogenital tract in females. In a typical cloaca, the urethra, vagina, and rectum converge inside the pelvis, creating a common channel that opens into a single orifice where the urethra is normally positioned. On the other hand, a posterior cloaca is atypical that the urogenital sinus runs posteriorly and opens between the anterior rectal wall and the anus.[9] In the discussion of absent penis, Penã et al. pointed out the noteworthy similarity to the anatomy of a posterior cloaca in female patients.[3] Therefore, this condition could be called “male posterior

cloaca". In the meanwhile, the URSM includes the absence of the perineal and anal openings in association with ambiguous genitalia and urogenital, colonic, and lumbosacral anomalies. The URSMS has been considered to be caused by a defect in cloacal differentiation during early embryogenesis. URSMS is classified into two conditions, "partial URSMS" or "full URSMS". Partial URSMS has characteristics of a single perineal or anal opening draining a common cloaca associated with a variety of unusual external genital abnormalities, whereas full URSMS has characteristics of the absence of both perineal and anal openings.[7,8] Some authors think aphallia falls into the category of URSMS.[10]

However, what we should call a condition is not an essential issue but an incidental detail in clinical practice. Different perspectives lead to different views of the same condition. In particular, development of the genital tubercle and induction of cloacal differentiation are thought to occur at the same time, which explains why urogenital anomalies are often associated with ARM. [10]

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In conclusion, the wide spectrum of urogenital abnormalities and ARM sometimes makes it difficult to name the conditions simply. However, more important than naming them is understanding the clinical conditions precisely.

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